

# SEQUENCE LISTING

<110> Dean, Michael  
Allikmets, Rando  
Hutchinson, Amy A.

<120> ATP-BINDING TRANSPORTER (ABC7) AND METHODS FOR DETECTION OF ANEMIA AND ATAXIA

<130> 4239-67289

<150> US 09/422,840

<151> 1999-10-21

<150> US 60/105,497

<151> 1998-10-23

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<170> PatentIn version 3.1

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<212> DNA

<213> Homo sapien

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gtcaatagtg tgaaaggctg tggaaactgt tcgtgctaag tcacataaga cattttcttt	2280
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 Gly Pro Gln Trp Arg Pro His Gln Leu Gly Ala Leu Gly Thr Ala Arg  
 35 40 45  
 Ala Tyr Gln Ile Pro Glu Ser Leu Lys Ser Ile Thr Trp Gln Arg Leu  
 50 55 60  
 Gly Lys Gly Asn Ser Gly Gln Phe Leu Asp Ala Ala Lys Ala Leu Gln  
 65 70 75 80  
 Val Trp Pro Leu Ile Glu Lys Arg Thr Cys Trp His Gly His Ala Gly  
 85 90 95  
 Gly Gly Leu His Thr Asp Pro Lys Glu Gly Leu Lys Asp Val Asp Thr  
 100 105 110  
 Arg Lys Ile Ile Lys Ala Met Leu Ser Tyr Val Trp Pro Lys Asp Arg  
 115 120 125  
 Pro Asp Leu Arg Ala Arg Val Ala Ile Ser Leu Gly Phe Leu Gly Gly  
 130 135 140  
 Ala Lys Ala Met Asn Ile Val Val Pro Phe Met Phe Lys Tyr Ala Val  
 145 150 155 160  
 Asp Ser Leu Asn Gln Met Ser Gly Asn Met Leu Asn Leu Ser Asp Ala  
 165 170 175  
 Pro Asn Thr Val Ala Thr Met Ala Thr Ala Val Leu Ile Gly Tyr Gly  
 180 185 190  
 Val Ser Arg Ala Gly Ala Ala Phe Phe Asn Glu Val Arg Asn Ala Val  
 195 200 205  
 Phe Gly Lys Val Ala Gln Asn Ser Ile Arg Arg Ile Ala Lys Asn Val  
 210 215 220

Phe Leu His Leu His Asn Leu Asp Leu Gly Phe His Leu Ser Arg Gln  
 225 230 235 240

Thr Gly Ala Leu Ser Lys Ala Ile Asp Arg Gly Thr Lys Gly Ile Ser  
 245 250 255

Phe Val Leu Ser Ala Leu Val Phe Asn Leu Leu Pro Ile Met Phe Glu  
 260 265 270

Val Met Leu Val Ser Gly Val Leu Tyr Tyr Lys Cys Gly Ala Gln Phe  
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Ala Leu Val Thr Leu Gly Thr Leu Gly Thr Tyr Thr Ala Phe Thr Val  
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Ala Val Thr Arg Trp Arg Thr Arg Phe Arg Ile Glu Met Asn Lys Ala  
 305 310 315 320

Asp Asn Asp Ala Gly Asn Ala Ala Ile Asp Ser Leu Leu Asn Tyr Glu  
 325 330 335

Thr Val Lys Tyr Phe Asn Asn Glu Arg Tyr Glu Ala Gln Arg Tyr Asp  
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Gly Phe Leu Lys Thr Tyr Glu Thr Ala Ser Leu Lys Ser Thr Ser Thr  
 355 360 365

Leu Ala Met Leu Asn Phe Gly Gln Ser Ala Ile Phe Ser Val Gly Leu  
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Thr Ala Ile Met Val Leu Ala Ser Gln Gly Ile Val Ala Gly Thr Leu  
 385 390 395 400

Thr Val Gly Asp Leu Val Met Val Asn Gly Leu Leu Phe Gln Leu Ser  
 405 410 415

Leu Pro Leu Asn Phe Leu Gly Thr Val Tyr Arg Glu Thr Arg Gln Ala  
 420 425 430

Leu Ile Asp Met Asn Thr Leu Phe Thr Leu Leu Lys Val Asp Thr Gln  
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Ile Lys Asp Lys Val Met Ala Ser Pro Leu Gln Ile Thr Pro Gln Thr

450		455		460
Ala Thr Val Ala Phe Asp Asn Val His Phe Glu Tyr Ile Glu Gly Gln				
465		470		475 480
Lys Val Leu Ser Gly Ile Ser Phe Glu Val Pro Ala Gly Lys Lys Val				
	485		490	495
Ala Ile Val Gly Gly Ser Gly Ser Gly Lys Ser Thr Ile Val Arg Leu				
	500		505	510
Leu Phe Arg Phe Tyr Glu Pro Gln Lys Gly Ser Ile Tyr Leu Ala Gly				
	515		520	525
Gln Asn Ile Gln Asp Val Ser Leu Glu Ser Leu Arg Arg Ala Val Gly				
	530		535	540
Val Val Pro Gln Asp Ala Val Leu Phe His Asn Thr Ile Tyr Tyr Asn				
545		550		555 560
Leu Leu Tyr Gly Asn Ile Ser Ala Ser Pro Glu Glu Val Tyr Ala Val				
	565		570	575
Ala Lys Leu Ala Gly Leu His Asp Ala Ile Leu Arg Met Pro His Gly				
	580		585	590
Tyr Asp Thr Gln Val Gly Glu Arg Gly Leu Lys Leu Ser Gly Gly Glu				
	595		600	605
Lys Gln Arg Val Ala Ile Ala Arg Ala Ile Leu Lys Asp Pro Pro Val				
	610		615	620
Ile Leu Tyr Asp Glu Ala Thr Ser Ser Leu Asp Ser Ile Thr Glu Glu				
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Thr Ile Leu Gly Ala Met Lys Asp Val Val Lys His Arg Thr Ser Ile				
	645		650	655
Phe Ile Ala His Arg Leu Ser Thr Val Val Asp Ala Asp Glu Ile Ile				
	660		665	670
Val Leu Asp Gln Gly Lys Val Ala Glu Arg Gly Thr His His Gly Leu				
	675		680	685

Leu Ala Asn Pro His Ser Ile Tyr Ser Glu Met Trp His Thr Gln Ser  
 690 695 700

Ser Arg Val Gln Asn His Asp Asn Pro Lys Trp Glu Ala Lys Lys Glu  
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Asn Ile Ser Lys Glu Glu Glu Arg Lys Lys Leu Gln Glu Glu Ile Val  
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Asn Ser Val Lys Gly Cys Gly Asn Cys Ser Cys  
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Gly Pro Gln Trp Arg Pro His Gln Leu Gly Ala Leu Gly Thr Ala Arg  
35 40 45

Ala Tyr Gln Ile Pro Glu Ser Leu Lys Ser Ile Thr Trp Gln Arg Leu  
50 55 60

Gly Lys Gly Asn Ser Gly Gln Phe Leu Asp Ala Ala Lys Ala Leu Gln  
65 70 75 80

Val Trp Pro Leu Ile Glu Lys Arg Thr Cys Trp His Gly His Ala Gly  
85 90 95

Gly Gly Leu His Thr Asp Pro Lys Glu Gly Leu Lys Asp Val Asp Thr  
100 105 110

Arg Lys Ile Ile Lys Ala Met Leu Ser Tyr Val Trp Pro Lys Asp Arg  
115 120 125

Pro Asp Leu Arg Ala Arg Val Ala Ile Ser Leu Gly Phe Leu Gly Gly  
130 135 140

Ala Lys Ala Met Asn Ile Val Val Pro Phe Met Phe Lys Tyr Ala Val  
145 150 155 160

Asp Ser Leu Asn Gln Met Ser Gly Asn Met Leu Asn Leu Ser Asp Ala  
165 170 175

Pro Asn Thr Val Ala Thr Met Ala Thr Ala Val Leu Ile Gly Tyr Gly  
180 185 190

Val Ser Arg Ala Gly Ala Ala Phe Phe Asn Glu Val Arg Asn Ala Val  
195 200 205

Phe Gly Lys Val Ala Gln Asn Ser Ile Arg Arg Ile Ala Lys Asn Val  
210 215 220

Phe Leu His Leu His Asn Leu Asp Leu Gly Phe His Leu Ser Arg Gln  
225 230 235 240



Thr Gly Ala Leu Ser Lys Ala Ile Asp Arg Gly Thr Lys Gly Ile Ser  
245 250 255

Phe Val Leu Ser Ala Leu Val Phe Asn Leu Leu Pro Ile Met Phe Glu  
260 265 270

Val Met Leu Val Ser Gly Val Leu Tyr Tyr Lys Cys Gly Ala Gln Phe  
275 280 285

Ala Leu Val Thr Leu Gly Thr Leu Gly Thr Tyr Thr Ala Phe Thr Val  
290 295 300

Ala Val Thr Arg Trp Arg Thr Arg Phe Arg Ile Glu Met Asn Lys Ala  
305 310 315 320

Asp Asn Asp Ala Gly Asn Ala Ala Ile Asp Ser Leu Leu Asn Tyr Glu  
325 330 335

Thr Val Lys Tyr Phe Asn Asn Glu Arg Tyr Glu Ala Gln Arg Tyr Asp  
340 345 350

Gly Phe Leu Lys Thr Tyr Glu Thr Ala Ser Leu Lys Ser Thr Ser Thr  
355 360 365

Leu Ala Met Leu Asn Phe Gly Gln Ser Ala Ile Phe Ser Val Gly Leu  
370 375 380

Thr Ala Ile Met Val Leu Ala Ser Gln Gly Met Val Ala Gly Thr Leu  
385 390 395 400

Thr Val Gly Asp Leu Val Met Val Asn Gly Leu Leu Phe Gln Leu Ser  
405 410 415

Leu Pro Leu Asn Phe Leu Gly Thr Val Tyr Arg Glu Thr Arg Gln Ala  
420 425 430

Leu Ile Asp Met Asn Thr Leu Phe Thr Leu Leu Lys Val Asp Thr Gln  
435 440 445

Ile Lys Asp Lys Val Met Ala Ser Pro Leu Gln Ile Thr Pro Gln Thr  
450 455 460

Ala Thr Val Ala Phe Asp Asn Val His Phe Glu Tyr Ile Glu Gly Gln  
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Lys Val Leu Ser Gly Ile Ser Phe Glu Val Pro Ala Gly Lys Lys Val  
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Ala Ile Val Gly Gly Ser Gly Ser Gly Lys Ser Thr Ile Val Arg Leu  
 500 505 510

Leu Phe Arg Phe Tyr Glu Pro Gln Lys Gly Ser Ile Tyr Leu Ala Gly  
 515 520 525

Gln Asn Ile Gln Asp Val Ser Leu Glu Ser Leu Arg Arg Ala Val Gly  
 530 535 540

Val Val Pro Gln Asp Ala Val Leu Phe His Asn Thr Ile Tyr Tyr Asn  
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Leu Leu Tyr Gly Asn Ile Ser Ala Ser Pro Glu Glu Val Tyr Ala Val  
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Ala Lys Leu Ala Gly Leu His Asp Ala Ile Leu Arg Met Pro His Gly  
 580 585 590

Tyr Asp Thr Gln Val Gly Glu Arg Gly Leu Lys Leu Ser Gly Gly Glu  
 595 600 605

Lys Gln Arg Val Ala Ile Ala Arg Ala Ile Leu Lys Asp Pro Pro Val  
 610 615 620

Ile Leu Tyr Asp Glu Ala Thr Ser Ser Leu Asp Ser Ile Thr Glu Glu  
 625 630 635 640

Thr Ile Leu Gly Ala Met Lys Asp Val Val Lys His Arg Thr Ser Ile  
 645 650 655

Phe Ile Ala His Arg Leu Ser Thr Val Val Asp Ala Asp Glu Ile Ile  
 660 665 670

Val Leu Asp Gln Gly Lys Val Ala Glu Arg Gly Thr His His Gly Leu  
 675 680 685

Leu Ala Asn Pro His Ser Ile Tyr Ser Glu Met Trp His Thr Gln Ser

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 Ser Arg Val Gln Asn His Asp Asn Pro Lys Trp Glu Ala Lys Lys Glu  
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 Asn Ile Ser Lys Glu Glu Glu Arg Lys Lys Leu Gln Glu Glu Ile Val  
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                                     35                                      40                                      45  
 Asn Leu Trp Asn Gln Ala Gln Lys Asp Ile Thr His Lys Lys Ser Val  
                                     50                                      55                                      60  
 Glu Gln Phe Ser Ser Ala Pro Lys Val Lys Thr Gln Val Lys Lys Thr  
 65                                      70                                      75                                      80  
 Ser Lys Ala Pro Thr Leu Ser Glu Leu Lys Ile Leu Lys Asp Leu Phe  
                                     85                                      90                                      95

Arg Tyr Ile Trp Pro Lys Gly Asn Asn Lys Val Arg Ile Arg Val Leu  
 100 105 110

Ile Ala Leu Gly Leu Leu Ile Ser Ala Lys Ile Leu Asn Val Gln Val  
 115 120 125

Pro Phe Phe Phe Lys Gln Thr Ile Asp Ser Met Asn Ile Ala Trp Asp  
 130 135 140

Asp Pro Thr Val Ala Leu Pro Ala Ala Ile Gly Leu Thr Ile Leu Cys  
 145 150 155 160

Tyr Gly Val Ala Arg Phe Gly Ser Val Leu Phe Gly Glu Leu Arg Asn  
 165 170 175

Ala Val Phe Ala Lys Val Ala Gln Asn Ala Ile Arg Thr Val Ser Leu  
 180 185 190

Gln Thr Phe Gln His Leu Met Lys Leu Asp Leu Gly Trp His Leu Ser  
 195 200 205

Arg Gln Thr Gly Gly Leu Thr Arg Ala Met Asp Arg Gly Thr Lys Gly  
 210 215 220

Ile Ser Gln Val Leu Thr Ala Met Val Phe His Ile Ile Pro Ile Ser  
 225 230 235 240

Phe Glu Ile Ser Val Val Cys Gly Ile Leu Thr Tyr Gln Phe Gly Ala  
 245 250 255

Ser Phe Ala Ala Ile Thr Phe Ser Thr Met Leu Leu Tyr Ser Ile Phe  
 260 265 270

Thr Ile Lys Thr Thr Ala Trp Arg Thr His Phe Arg Arg Asp Ala Asn  
 275 280 285

Lys Ala Asp Asn Lys Ala Ala Ser Val Ala Leu Asp Ser Leu Ile Asn  
 290 295 300

Phe Glu Ala Val Lys Tyr Phe Asn Asn Glu Lys Tyr Leu Ala Asp Lys  
 305 310 315 320

Tyr Asn Gly Ser Leu Met Asn Tyr Arg Asp Ser Gln Ile Lys Val Ser  
325 330 335

Gln Ser Leu Ala Phe Leu Asn Ser Gly Gln Asn Leu Ile Phe Thr Thr  
340 345 350

Ala Leu Thr Ala Met Met Tyr Met Gly Cys Thr Gly Val Ile Gly Gly  
355 360 365

Asn Leu Thr Val Gly Asp Leu Val Leu Ile Asn Gln Leu Val Phe Gln  
370 375 380

Leu Ser Val Pro Leu Asn Phe Leu Gly Ser Val Tyr Arg Asp Leu Lys  
385 390 395 400

Gln Ser Leu Ile Asp Met Glu Thr Leu Phe Lys Leu Arg Lys Asn Glu  
405 410 415

Val Lys Ile Lys Asn Ala Glu Arg Pro Leu Met Leu Pro Glu Asn Val  
420 425 430

Pro Tyr Asp Ile Thr Phe Glu Asn Val Thr Phe Gly Tyr His Pro Asp  
435 440 445

Arg Lys Ile Leu Lys Asn Ala Ser Phe Thr Ile Pro Ala Gly Trp Lys  
450 455 460

Thr Ala Ile Val Gly Ser Ser Gly Ser Gly Lys Ser Thr Ile Leu Lys  
465 470 475 480

Leu Val Phe Arg Phe Tyr Asp Pro Glu Ser Gly Arg Ile Leu Ile Asn  
485 490 495

Gly Arg Asp Ile Lys Glu Tyr Asp Ile Asp Ala Leu Arg Lys Val Ile  
500 505 510

Gly Val Val Pro Gln Asp Thr Pro Leu Phe Asn Asp Thr Ile Trp Glu  
515 520 525

Asn Val Lys Phe Gly Arg Ile Asp Ala Thr Asp Glu Glu Val Ile Thr  
530 535 540

Val Val Glu Lys Ala Gln Leu Ala Pro Leu Ile Lys Lys Leu Pro Gln

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Ile	Met	Phe	Phe	Asp	Glu	Ala	Thr	Ser	Ala	Leu	Asp	Thr	His	Thr	Glu
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Gln	Ala	Leu	Leu	Arg	Thr	Ile	Arg	Asp	Asn	Phe	Thr	Ser	Gly	Ser	Arg
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Glu	Leu														
	690														